

WE CLAIM:

1. A method for detecting the presence in a subject of a polymorphism linked to a gene associated with familial dysautonomia which comprises:

analyzing human chromosome 9 of the subject and detecting the presence of a polymorphism located between D9S53 and D9S105 inclusive and linked to the gene associated with familial dysautonomia wherein the presence of the polymorphism is indicative of carriers of a gene associated with familial dysautonomia.

2. The method according to claim 1, wherein the polymorphism is selected from the group consisting of D9S53, D9S310, D9S309, D9S172 and D9S174.

3. A method for detecting the presence of polymorphisms linked to a gene associated with familial dysautonomia in a subject comprising detecting a maternal polymorphism linked to a gene associated with familial dysautonomia; detecting a paternal polymorphism linked to a gene associated with familial dysautonomia; typing the subject to determine the maternal polymorphism and paternal polymorphism; linking the distribution of the maternal polymorphism and paternal polymorphism with familial dysautonomia; and determining if the subject has the polymorphism located on the long arm human chromosome 9

between D9S53 and D9S105 inclusive linked with a gene associated with familial dysautonomia.

4. The method according to claim 1 wherein the analysis includes amplifying the polymorphism.

5. The method according to claim 1 wherein the polymorphism is detected by autoradiography.

6. The method according to claim 1 wherein the analysis includes comparison of the polymorphism of the subject to the polymorphism of each parent of the subject which are unaffected by familial dysautonomia disease and a family member affected by familial dysautonomia disease.

7. A method for detecting the presence of a polymorphism linked to a gene associated with familial dysautonomia in a subject comprising typing blood relatives of a subject for a polymorphism located on the long arm of human chromosome 9 between D9S53 and D9S105 inclusive linked with a gene associated with the familial dysautonomia; and analyzing DNA from the subject for the presence of the polymorphism.

8. An isolated and purified polynucleotide sequence comprising a gene associated with familial

dysautonomia and wherein the polynucleotide sequence is isolated from the 9q31-q33 region of chromosome 9.

5 9. An isolated and purified polynucleotide sequence comprising a gene associated with familial dysautonomia and wherein the polynucleotide sequence is isolated from the long arm of chromosome 9 between D9S127
10 and D9S59.

10 10. The isolated and purified polynucleotide sequence according to claim 8 wherein the sequence consists
15 essentially of the sequence located between D9S53 and D9S105.

20 11. A kit for detecting the presence of polymorphisms associated with the gene for familial dysautonomia in an individual, the kit comprising at least one nucleic acid probe having a sequence that can identify
25 a polymorphism located between HXB and D9S109 inclusive and linked to the familial dysautonomia gene.

30 12. The kit according to claim 11 wherein the nucleic acid probe can identify a polymorphism selected from the group consisting of D9S53 (SEQ ID NOS: 15 and 16), D9S58 (SEQ ID NOS: 5 and 6), D9S59 (SEQ ID NOS: 7 and 8), D9S127 (SEQ ID NOS: 3 and 4), HXB (SEQ ID NOS: 1 and 2), D9S109
35 (SEQ ID NOS: 19 and 20), D9S106 (SEQ ID NOS: 21 and 22) and

° D9S105 (SEQ ID NOS: 17 and 18), D9S309, D9S310, D9S172 and D9S174.

5 13. An isolated nucleic acid sequence comprising about 15 to about 40 base pairs, wherein said sequence encodes the region flanking the DNA containing the D9S309 or D9S310 polymorphism.

10 14. An isolated nucleic acid sequence comprising the nucleic acid sequence 5' GCCTGGGCAAACAGAGAC3' or 5' GCAACTTATTGTTTAACTG-3'.

15 15. An isolated nucleic acid sequence comprising the nucleic acid sequence 5'-TAGAGCTCTACCCCCAAC-3' or 5'-TGAACAGCTATATATGCCATCC-3'.

20 16. An isolated nucleic acid sequence comprising the nucleic acid sequence 5' GCCTGGGCAAACAGAGAC3' or 5' GCAACTTATTGTTTAACTG-3'.

25 17. An isolated nucleic acid sequence containing the nucleic acid sequence 5'-TAGAGCTCTACCCCCAAC-3' or 5'-TGAACAGCTATATATGCCATCC-3'.

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